CLARIFY

medical clinicians
sensitive to light
susceptible

FORMS OF GENETIC DISORDER

As the scientific world learns more about the different forms of genetic disorder, the discovered information is recorded. **It can then help medical clinicians to diagnose and devise treatments or strategies for living with a disorder.** Those disorders include:

- **Albinism**
- **Ectrodactyly**
- **Hypertrichosis**
- **Intersex (hermaphroditism)**
- **Down syndrome**
- **Colour blindness**
- **Heterochromia**
- **Haemophilia**
- **Conjoined twins**
- **Progeria**
- **Dwarfism**

ALBINISM

Albinism is one of the most widely recognised genetic conditions. It makes a person unable to produce melanin, a pigment that gives colour to hair, skin and the iris of the eye. There are two major forms: oculocutaneous albinism, in which the affected person has white or pink hair, skin and iris colour, as well as vision problems; and ocular albinism, which only affects the eyes.

RESEARCH

Is albinism common to all nationalities? Find out.

People with albinism are sometimes sensitive to light, or have crossed eyes or vision problems. **Eye problems occur because the eye requires pigment in order to develop normal vision.** Often, surgery can help eye problems associated with albinism.

Although it's a physically striking condition, albinism is not very uncommon. Some form of it occurs in approximately one in 17,000 human births. It also appears in plants, insects, fish, reptiles, amphibians, birds, marsupials and mammals throughout the world.

The gene for albinism is recessive, which means that a person must inherit two copies of it – one from each parent – to show signs of the disorder. If the person inherits one gene for albinism and one normal gene, the normal gene will dominate. It will have enough genetic information to produce the pigment.

Though albinism does not affect a person's lifespan, it might limit their choice of outdoor activities. Those with albinism are highly sensitive to sun and susceptible to skin cancer, so they must be extra cautious to avoid exposure.

VISUAL FEATURES

How could you visually interpret the text explaining how people can inherit albinism from their parents or just miss being affected by it?

CLARIFY

malformed
manifest
digit
syndrome
foetus

ECTRODACTYLY

The name ectrodactyly is derived from the Greek words *extroma* (meaning "abortion", or "termination of development") and *daktylos* (meaning "finger"). Ectrodactyly is a condition that involves missing or malformed fingers or toes.

This genetic condition can manifest itself in several ways. Sometimes it affects just one digit; other times it produces a cleft hand or foot, where the central digit is missing and the remaining ones on either side are webbed together. Once known as "lobster claw syndrome", this second form of ectrodactyly affects approximately one in every 55,000 to 70,000 people.

Like many genetic disorders, ectrodactyly is congenital, meaning that it's acquired during the development of the foetus and present at birth. Scientists have identified a number of genes connected with the disorder, and their research has given us a better understanding of the people who have it. In the past, ectrodactyly patients might have been hidden away or exhibited in circus shows. These days, they are getting more involved in educating the public about their condition.

QUESTION

What adaptations in the home or workplace might support people with ectrodactyly and minimise the effect of their disorder?

PERSONAL RESPONSE

Do the terms lobster claw syndrome and ectrodactyly evoke a different response? How did these terms affect your perception of this disorder?

...IN THE PAST, ECTRODACTYLY PATIENTS MIGHT HAVE BEEN HIDDEN AWAY OR EXHIBITED IN CIRCUS SHOWS

ISSUES

What issues arise with ectrodactyly patients being hidden away? What is your stance on how people with this genetic condition were treated?